A Case with Partial 9p Trisomy and Speech Impairment

Parsiyel 9p Trizomili Bir Olgu ve Konuşma Bozukluğu

9p Trizomisi ve Konuşma Bozukluğu / 9p Trisomy and Speech İmpairment

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Özet

Bu çalışmada, sitogenetik analiz amacıyla laboratuarımıza refere edilen 5 yaşında bir erkek çocukta saptanan der(15), ish t(9;15)(p12;p11,2) translokasyon ve olgunun fenotipik anomalileri arasındaki ilişkileri tartışmayı amaçladık. 5 yaşındaki erkek olgumuz konuşma bozukluğu ve gelişme geriliği şikayetlerine sahipken, olgunun aile bireylerinde herhangi bir fenotipik anomali bulunmamaktaydı. Olgunun aile öyküsünün değerlendirilmesinin ardından, periferik kan kültürü metodu kullanarak olgu ve aile bireylerinden elde edilen preparatlar GTG bantlama yöntemiyle değerlendirildi. Bunun yanında, olgunun annesi ve babasında yapılan sitogenetik analizler normal karyotiplere sahip olduklarını gösterdi. Olgumuzun sahip olduğu translokasyonun, bilinmeyen de novo mekanizmalar sonucu ortaya çıktığı kanaati edinildi. Bu çalışmada, olgunun fenotipik anomalileri ve genetik özellikleri arasındaki ilişkileri literatür ışığında tartıştık.

Anahtar Kelimeler

Kromozomal Anomali; Parsiyel Trizomi; Konuşma Bozukluğu

Abstract

In this study, we aimed to discuss the relationships between his phenotypic anomalies and der(15), ish t(9;15)(p12;q10) balanced reciprocal translocation entity that determined from 5 year old male child who referred to our laboratory for cytogenetic analysis. He has complaints speech impairment and growth retardation. And whose family has no phenotypic anomalies. After assessing the case's pedigree, the preparations obtained from case and relatives using peripheric blood culture method and evaluated with GTG banding. Cytogenetic analyses of her father and mother revealed normal karyotypes. We convinced that the balanced reciprocal translocation may be as a result of de novo mechanism. So, we discussed relationship between phenotypic anomalies and genetic characteristics of case in the light of the literature.

Keywords

Chromosomal Anomaly; Partial Trisomy; Speech Impairment

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Introduction

Humans are characterized by an extraordinarily complex system of their verbal communication. There are two types of human communication including speech and language. Speech generally defines to the mechanical direction of verbal communication. Making sentences, sentence defining with sound, fluency are necessary qualifications for speech. Language is a complex function that are making meaningful words within the rules, making new words, using together of words, allowing the construction of word combinations in specific situations. The barriers of human communication is categorized as language and speech impairment. Speech impairments are include sentence building problems, speech fluency problems and dyspraxia [1]. Developmental speech and language disorders play an important role within childhood diseases in spite of different phenotypes based on heterogeneous factors. Speech and language development disorders is identified five categories [2]. These include mixed receptive-expressive language disorder, expressive language disorder, phonological disorder, stuttering and communication disorders [3]. The most of language and speech disorders are seen to accompanied other pathological features in spite of recognized as clinically. While the approximately 15% of children with persistent speech impairment have development a language disorders. Approximately %5 of children with speech and language disorders have speech difficulty at the same time [3,4].

Trisomy 9p is one of the autosomal aneuploidies that has long survival. The clinical findings in patients with trisomy 9p is directly related to size of duplicated chromosomal material. Trisomy 9p syndrome is defined to clinically and the most significant finding of it's is craniofacial dysmorphism [5]. In addition, it is reported that neurological problems, speech and language disorders are seen in cases with trisomy 9p [6-8].

Case Report

Cytogenetic analysis of case, who referred to our laboratory with speech impairment (such as verbal dyspraxia and speech fluency problems) and growth retardation, showed that has been a carrier of balanced reciprocal translocation (46,XY, der(15), ish t(9;15)(p12;q11.2) (Figure 1, 2). Cytogenetic analyses of his parents, two sisters and two brothers revealed normal karyotypes (Figure 3). Karyotype analyses of cases were made from their peripheral blood lymphocytes by standard method. Fifty metaphases had been prepared with GTG banding method for each patient was analyzed. Thirty metaphases were karyotyped. Karyotypes were described by using ISCN 2005 standard nomenclature of human chromosomes. In addition, fluorescence in-situ hybridization (FISH) analysis was performed the case.

Discussion

Genetic factors were determined to have effects on formation of language and speech abnormalities. By the studies searching the genetic factors that take a part in the formation of language and speech abnormalities; ıt was understood that the formation of language and speech abnormalities in both of the twins were more common in monozygotic twins than dizygotic twins. The reason of the appearance of language and speech abnormalities in monozygotic twins more commonly than dizygotic twins was reported to origin from familial accumulation in addition to similar genetic factors in monozygotic twins [9-11]. Environmental factors interacting with genetic factors were shown to have influence on the formation of language and speech abnormalities, so, it has a multifactorial inheritance [3,12,13]. Although genetic factors were shown to have effect on language and speech abnormalities, its pathology has not been understood yet [3].

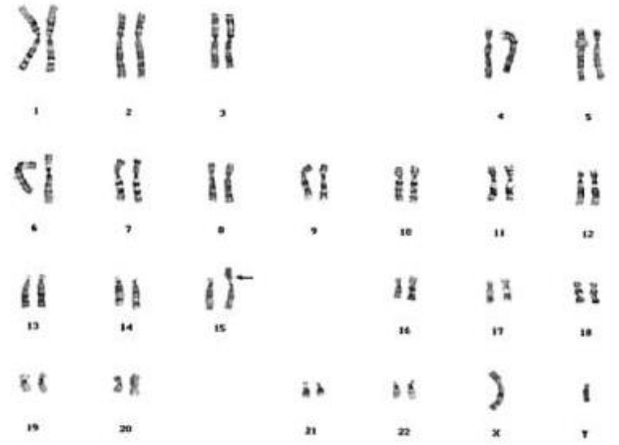

Figure 1. Karyotype of the case der(15), t(9;15)(p12;p11,2)

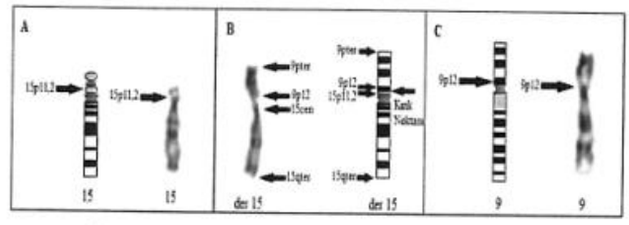

Figure 2. Chromosome 9 and 15 ideograms of the cases with GTG band (A-C) Ideogram (B)

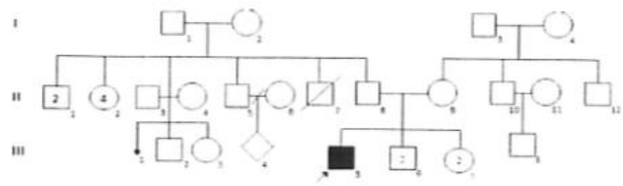

Figure 3. Pedigre of the case

In a report published by Rossi et al., after the evaluation of the case with delayed development, microcephaly, micrognathy, brachycephalic, bulbous nose, downturned oral commissures, malformed ears and feet, and hypotonia; one of the parents of the case was estimated to be carrier of t(9;15), so the case had language and speech abnormalities because of partial trisomy 9p [8]. In another report published by de Pater et al., similar findings with Rossi et al. were observed in a partial trisomy 9p case whose mother was carrier of t(9;12) [6]. By another study which was reported by Hauge et al. with 10 cases of 9p deletion, it was reported that there were genes associated with language and speech development, neurologic development, so language and speech delay may occur as result of copy number variations in these regions [7]. The results of Rossi et al., de Pater et al. and Hauge et al., presented that various regions in p arm of the chromosome 9 carry important genes for the development of language and speech and it was understood that the findings of our case were in concordance with the literature reports. As

a result, our examinations showed that partial 9p trisomy der (15) t(9;15)(p12;p11,2) karyotype occured de novo. Though the formation mechanisms of the translocations are not exactly known. It is believed that the recombination is basic mechanism. Phenotypical findings of this patient with partial trisomy 9p discussed with other trisomy 9p cases in the literature. In the present case speech disorders were observed as a result of 9p trisomy. To protect the health of future generations; importance of prenatal diagnosis and genetic counseling were emphasized in this study.

Competing interests

The authors declare that they have no competing interests.

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